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HELSINGFORS UNIVERSITET
UNIVERSITY OF HELSINKI

INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI 2005–2010

RC-Specific Evaluation of MUSGEN – Gene-culture evolution in music

Seppo Saari & Antti Moilanen (Eds.)

Evaluation Panel: Biological, Agricultural and Veterinary Sciences

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University of Helsinki
Administrative Publications 80/8
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Title:
International Evaluation of Research and Doctoral Training at the University of Helsinki 2005–2010 : RC-Specific Evaluation of MUSGEN – Gene-culture evolution in music

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Summary:
Researcher Community (RC) was a new concept of the participating unit in the evaluation. Participation in the evaluation was voluntary and the RCs had to choose one of the five characteristic categories to participate.
Evaluation of the Researcher Community was based on the answers to the evaluation questions. In addition a list of publications and other activities were provided by the TUHAT system. The CWTS/Leiden University conducted analyses for 80 RCs and the Helsinki University Library for 66 RCs.
Panellists, 49 and two special experts in five panels evaluated all the evaluation material as a whole and discussed the feedback for RC-specific reports in the panel meetings in Helsinki. The main part of this report is consisted of the feedback which is published as such in the report.
Chapters in the report:
1. Background for the evaluation
2. Evaluation feedback for the Researcher Community
3. List of publications
4. List of activities
5. Bibliometric analyses
The level of the RCs' success can be concluded from the written feedback together with the numeric evaluation of four evaluation questions and the category fitness. More conclusions of the success can be drawn based on the University-level report.

RC-specific information:

Main scientific field of research:
Biological, Agricultural and Veterinary Sciences

RC-specific keywords:
gene, culture, SNP, association, evolution, musical aptitude, environment, education, interaction, expression

Participation category:
4. Research of the participating community represents an innovative opening

RC's responsible person:
Järvelä, Irma

Keywords:
Research Evaluation, Meta-evaluation, Doctoral Training, Bibliometric Analyses, Researcher Community

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Contents

Foreword

The evaluation of research and doctoral training is being carried out in the years 2010–2012 and will end in 2012. The steering group appointed by the Rector in January 2010 set the conditions for participating in the evaluation and prepared the Terms of Reference to present the evaluation procedure and criteria. The publications and other scientific activities included in the evaluation covered the years 2005–2010.

The participating unit in the evaluation was defined as a Researcher Community (RC). To obtain a critical mass with university-level impact, the number of members was set to range from 20 to 120. The RCs were required to contain researchers in all stages of their research career, from doctoral students to principal investigators (PIs). All in all, 136 Researcher Communities participated in this voluntary evaluation, 5857 persons in total, of whom 1131 were principal investigators. PIs were allowed to participate in two communities in certain cases, and 72 of them used this opportunity and participated in two RCs.

This evaluation enabled researchers to define RCs from the "bottom up" and across disciplines. The aim of the evaluation was not to assess individual performance but a community with shared aims and researcher-training activities. The RCs were able to choose among five different categories that characterised the status and main aims of their research. The steering group considered the process of applying to participate in the evaluation to be important, which lead to the establishment of these categories. In addition, providing a service for the RCs to enable them to benchmark their research at the global level was a main goal of the evaluation.

The data for the evaluation consisted of the RCs' answers to evaluation questions on supplied e-forms and a compilation extracted from the TUHAT – Research Information System (RIS) on 12 April 2011. The compilation covered scientific and other publications as well as certain areas of scientific activities. During the process, the RCs were asked to check the list of publications and other scientific activities and make corrections if needed. These TUHAT compilations are public and available on the evaluation project sites of each RC in the TUHAT-RIS.

In addition to the e-form and TUHAT compilation, University of Leiden (CWTS) carried out bibliometric analyses from the articles included in the Web of Science (WoS). This was done on University and RC levels. In cases where the publication forums of the RC were clearly not represented by the WoS data, the Library of the University of Helsinki conducted a separate analysis of the publications. This was done for 66 RCs representing the humanities and social sciences.

The evaluation office also carried out an enquiry targeted to the supervisors and PhD candidates about the organisation of doctoral studies at the University of Helsinki. This and other documents describing the University and the Finnish higher education system were provided to the panellists.

The panel feedback for each RC is unique and presented as an entity. The first collective evaluation reports available for the whole panel were prepared in July–August 2011. The reports were accessible to all panel members via the electronic evaluation platform in August. Scoring from 1 to 5 was used to complement written feedback in association with evaluation questions 1–4 (scientific focus and quality, doctoral training, societal impact, cooperation) and in addition to the category evaluating the fitness for participation in the evaluation. Panellists used the international level as a point of comparison in the evaluation. Scoring was not expected to go along with a preset deviation.

Each of the draft reports were discussed and dealt with by the panel in meetings in Helsinki (from 11 September to 13 September or from 18 September to 20 September 2011). In these meetings the panels also examined the deviations among the scores and finalised the draft reports together.

The current RC-specific report deals shortly with the background of the evaluation and the terms of participation. The main evaluation feedback is provided in the evaluation report, organised according to the evaluation questions. The original material provided by the RCs for the panellists has been attached to these documents.

On behalf of the evaluation steering group and office, I sincerely wish to thank you warmly for your participation in this evaluation. The effort you made in submitting the data to TUHAT-RIS is gratefully acknowledged by the University. We wish that you find this panel feedback useful in many ways. The bibliometric profiles may open a new view on your publication forums and provide a perspective for discussion on your choice of forums. We especially hope that this evaluation report will help you in setting the future goals of your research.

Johanna Björkroth
Vice-Rector
Chair of the Steering Group of the Evaluation

Steering Group of the evaluation

Steering group, nominated by the Rector of the University, was responsible for the planning of the evaluation and its implementation having altogether 22 meetings between February 2010 and March 2012.

Chair

Vice-Rector, professor **Johanna Björkroth**

Vice-Chair

Professor **Marja Airaksinen**

Chief Information Specialist, Dr **Maria Forsman**
Professor **Arto Mustajoki**
University Lecturer, Dr **Kirsi Pyhältö**
Director of Strategic Planning and Development, Dr **Ossi Tuomi**
Doctoral candidate, MSocSc **Jussi Vauhkonen**

Panel members

CHAIR
Professor Ary A. Hoffman
Ecological genetics, evolutionary biology, biodiversity conservation, zoology
University of Melbourne, Australia

VICE-CHAIR
Professor Barbara Koch
Forest Sciences, remote sensing
University of Freiburg, Germany

Professor Per-Anders Hansson
Agricultural engineering, modeling, life cycle analysis, bioenergy
Swedish University of Agricultural Sciences

Professor Danny Huylebroeck
Developmental biology
Katholieke Universiteit Leuven, Belgium

Professor Jonathan King
Virus assembly, protein folding
Massachusetts Institute of Technology MIT, USA

Professor Hannu J.T. Korhonen
Functional foods, dairy technology, milk hygiene
MTT Agrifood Research Finland

Professor Kristiina Kruus
Microbiological biotechnology, microbiological enzymes, applied microbiology
VTT Technical Research Centre of Finland

Professor Joakim Lundeberg
Biochemistry, biotechnology, sequencing, genomics
KTH Royal Institute of Technology, Sweden

Professor Dominiek Maes
Veterinary medicine
Ghent University, Belgium

Professor Olli Saastamoinen
Forest economics and policy
University of Eastern Finland

Professor Kai Simons
Biochemistry, molecular biology, cell biology
Max-Planck-Institute of Molecular Cell Biology and Genetics, Germany

The panel, independently, evaluated all the submitted material and was responsible for the feedback of the RC-specific reports. The panel members were asked to confirm whether they had any conflict of interests with the RCs. If this was the case, the panel members disqualified themselves in discussion and report writing.

Added expertise to the evaluation was contributed by the members from the other panels and by one evaluator outside the panels.

External Expert

Professor Anders Linde
Oral biochemi
Faculty of Odontology
Göteborg University
Sweden

Experts from the Other Panels

Professor Caitlin Buck, from the Panel of Natural Sciences
Professor Ritske Huismans, from the Panel of Natural Sciences
Professor Johanna Ivaska, from the Panel of Medicine, biomedicine and health sciences
Professor Lea Kauppi, from the Panel of Natural Sciences
Professor Holger Stark, from the Panel of Natural Sciences
Professor Peter York, from the Panel of Medicine, biomedicine and health sciences

EVALUATION OFFICE

Dr Seppo Saari, **Doc.**, Senior Adviser in Evaluation, was responsible for the entire evaluation, its planning and implementation and acted as an Editor-in-chief of the reports.

Dr Eeva Sievi, **Doc.,** Adviser, was responsible for the registration and evaluation material compilations for the panellists. She worked in the evaluation office from August 2010 to July 2011.

MSocSc Paula Ranne, Planning Officer, was responsible for organising the panel meetings and all the other practical issues like agreements and fees and editing a part the RC-specific reports. She worked in the evaluation office from March 2011 to January 2012.

Mr Antti Moilanen, Project Secretary, was responsible for editing the reports. He worked in the evaluation office from January 2012 to April 2012.

TUHAT OFFICE

Provision of the publication and other scientific activity data

Mrs Aija Kaitera, Project Manager of TUHAT-RIS served the project ex officio providing the evaluation project with the updated information from TUHAT-RIS. The TUHAT office assisted in mapping the publications with CWTS/University of Leiden.

MA Liisa Ekebom, Assisting Officer, served in TUHAT-RIS updating the publications for the evaluation. She also assisted the UH/Library analyses.

BA Liisa Jäppinen, Assisting Officer, served in TUHAT-RIS updating the publications for the evaluation.

HELSINKI UNIVERSITY LIBRARY

Provision of the publication analyses

Dr Maria Forsman, Chief Information Specialist in the Helsinki University Library, managed with her 10 colleagues the bibliometric analyses in humanities, social sciences and in other fields of sciences where CWTS analyses were not applicable.

Acronyms and abbreviations applied in the report

External competitive funding

AF – Academy of Finland
TEKES - Finnish Funding Agency for Technology and Innovation
EU - European Union
ERC - European Research Council
International and national foundations
FP7/6 etc. /Framework Programmes/Funding of European Commission

Evaluation marks

Outstanding (5)
Excellent (4)
Very Good (3)
Good (2)
Sufficient (1)

Abbreviations of Bibliometric Indicators

P - Number of publications
TCS – Total number of citations
MCS - Number of citations per publication, excluding self-citations
PNC - Percentage of uncited publications
MNCS - Field-normalized number of citations per publication
MNJS - Field-normalized average journal impact
THCP10 - Field-normalized proportion highly cited publications (top 10%)
INT_COV - Internal coverage, the average amount of references covered by the WoS
WoS – Thomson Reuters Web of Science Databases

Participation category

Category 1. The research of the participating community represents the international cutting edge in its field.
Category 2. The research of the participating community is of high quality, but the community in its present composition has yet to achieve strong international recognition or a clear break-through.
Category 3. The research of the participating community is distinct from mainstream research, and the special features of the research tradition in the field must be considered in the evaluation.
Category 4. The research of the participating community represents an innovative opening.
Category 5. The research of the participating community has a highly significant societal impact.

Research focus areas of the University of Helsinki

Focus area 1: The basic structure, materials and natural resources of the physical world
Focus area 2: The basic structure of life
Focus area 3: The changing environment – clean water
Focus area 4: The thinking and learning human being
Focus area 5: Welfare and safety
Focus area 6: Clinical research
Focus area 7: Precise reasoning
Focus area 8: Language and culture
Focus area 9: Social justice
Focus area 10: Globalisation and social change

1 Introduction to the Evaluation

1.1 RC-specific evaluation reports

The participants in the evaluation of research and doctoral training were Researcher Communities (hereafter referred to as the RC). The RC refers to the group of researchers who registered together in the evaluation of their research and doctoral training. Preconditions in forming RCs were stated in the Guidelines for the Participating Researcher Communities. The RCs defined themselves whether their compositions should be considered well-established or new.

It is essential to emphasise that the evaluation combines both meta-evaluation[1] and traditional research assessment exercise and its focus is both on the research outcomes and procedures associated with research and doctoral training. The approach to the evaluation is enhancement-led where self-evaluation constituted the main information. The answers to the evaluation questions formed together with the information of publications and other scientific activities an entity that was to be reviewed as a whole.

The present evaluation recognizes and justifies the diversity of research practices and publication traditions. Traditional Research Assessment Exercises do not necessarily value high quality research with low volumes or research distinct from mainstream research. It is challenging to expose the diversity of research to fair comparison. To understand the essence of different research practices and to do justice to their diversity was one of the main challenges of the present evaluation method. Understanding the divergent starting points of the RCs demanded sensitivity from the evaluators.

1.2 Aims and objectives in the evaluation

The aims of the evaluation are as follows:

- to improve the level of research and doctoral training at the University of Helsinki and to raise their international profile in accordance with the University's strategic policies. The improvement of doctoral training should be compared to the University's policy.[2]
- to enhance the research conducted at the University by taking into account the diversity, originality, multidisciplinary nature, success and field-specificity,
- to recognize the conditions and prerequisites under which excellent, original and high-impact research is carried out,
- to offer the academic community the opportunity to receive topical and versatile international peer feedback,
- to better recognize the University's research potential.
- to exploit the University's TUHAT research information system to enable transparency of publishing activities and in the production of reliable, comparable data.

1.3 Evaluation method

The evaluation can be considered as an enhancement-led evaluation. Instead of ranking, the main aim is to provide useful information for the enhancement of research and doctoral training of the participating RCs. The comparison should take into account each field of science and acknowledge their special character.

[1] The panellists did not read research reports or abstracts but instead, they evaluated answers to the evaluation questions, tables and compilations of publications, other scientific activities, bibliometrics or comparable analyses.

[2] Policies on doctoral degrees and other postgraduate degrees at the University of Helsinki.

The comparison produced information about the present status and factors that have lead to success. Also challenges in the operations and outcomes were recognized.

The evaluation approach has been designed to recognize better the significance and specific nature of researcher communities and research areas in the multidisciplinary top-level university. Furthermore, one of the aims of the evaluation is to bring to light those evaluation aspects that differ from the prevalent ones. Thus the views of various fields of research can be described and research arising from various starting points understood better. The doctoral training is integrated into the evaluation as a natural component related to research. Operational processes of doctoral training are being examined in the evaluation.

Five stages of the evaluation method were:

1. Registration – Stage 1
2. Self-evaluation – Stage 2
3. TUHAT[3] compilations on publications and other scientific activities[4]
4. External evaluation
5. Public reporting

1.4 Implementation of the external evaluation

Five Evaluation Panels

Five evaluation panels consisted of independent, renowned and highly respected experts. The main domains of the panels are:

1. biological, agricultural and veterinary sciences
2. medicine, biomedicine and health sciences
3. natural sciences
4. humanities
5. social sciences

The University invited 10 renowned scientists to act as chairs or vice-chairs of the five panels based on the suggestions of faculties and independent institutes. Besides leading the work of the panel, an additional role of the chairs was to discuss with other panel chairs in order to adopt a broadly similar approach. The panel chairs and vice-chairs had a pre-meeting on 27 May 2011 in Amsterdam.

The panel compositions were nominated by the Rector of the University 27 April 2011. The participating RCs suggested the panel members. The total number of panel members was 50. The reason for a smaller number of panellists as compared to the previous evaluations was the character of the evaluation as a meta-evaluation. The panellists did not read research reports or abstracts but instead, they evaluated answers to the evaluation questions, tables and compilations of publications, other scientific activities, bibliometrics and comparable analyses.

The panel meetings were held in Helsinki:

- On 11–13 September 2011: (1) biological, agricultural and veterinary sciences, (2) medicine, biomedicine and health sciences and (3) natural sciences.
- On 18–20 September 2011: (4) humanities and (5) social sciences.

[3] TUHAT (acronym) of Research Information System (RIS) of the University of Helsinki

[4] Supervision of thesis, prizes and awards, editorial work and peer reviews, participation in committees, boards and networks and public appearances.

1.5 Evaluation material

The main material in the evaluation was the RCs' self-evaluations that were qualitative in character and allowed the RCs to choose what was important to mention or emphasise and what was left unmentioned.

The present evaluation is exceptional at least in the Finnish context because it is based on both the evaluation documentation (self-evaluation questions, publications and other scientific activities) and the bibliometric reports. All documents were delivered to the panellists for examination.

Traditional bibliometrics can be reasonably done mainly in medicine, biosciences and natural sciences when using the Web of Science database, for example. Bibliometrics, provided by CWTS/The Centre for Science and Technology Studies, University of Leiden, cover only the publications that include WoS identification in the TUHAT-RIS.

Traditional bibliometrics are seldom relevant in humanities and social sciences because the international comparable databases do not store every type of high quality research publications, such as books and monographs and scientific journals in other languages than English. The Helsinki University Library has done analysis to the RCs, if their publications were not well represented in the Web of Science databases (RCs should have at least 50 publications and internal coverage of publications more than 40%) – it meant 58 RCs. The bibliometric material for the evaluation panels was available in June 2011. The RC-specific bibliometric reports are attached at the end of each report.

The panels were provided with the evaluation material and all other necessary background information, such as the basic information about the University of Helsinki and the Finnish higher education system.

Evaluation material

1. Registration documents of the RCs for the background information
2. Self evaluation material – answers to the evaluation questions
3. Publications and other scientific activities based on the TUHAT RIS:
   - 3.1. statistics of publications
   - 3.2. list of publications
   - 3.3. statistics of other scientific activities
   - 3.4. list of other scientific activities
4. Bibliometrics and comparable analyses:
   - 4.1. Analyses of publications based on the verification of TUHAT-RIS publications with the Web of Science publications (CWTS/University of Leiden)
   - 4.2. Publication statistics analysed by the Helsinki University Library - mainly for humanities and social sciences
5. University level survey on doctoral training (August 2011)
6. University level analysis on publications 2005–2010 (August 2011) provided by CWTS/University of Leiden

Background material

University of Helsinki
- Basic information about the University of the Helsinki
- The structure of doctoral training at the University of Helsinki
- Previous evaluations of research at the University of Helsinki – links to the reports: 1998 and 2005

The Finnish Universities/Research Institutes
- Finnish University system
- Evaluation of the Finnish National Innovation System
- The State and Quality of Scientific Research in Finland. Publication of the Academy of Finland 9/09.

The evaluation panels were provided also with other relevant material on request before the meetings in Helsinki.

1.6 Evaluation questions and material

The participating RCs answered the following evaluation questions which are presented according to the evaluation form. In addition, TUHAT RIS was used to provide the **additional material** as explained. For giving the feedback to the RCs, the panellists received the evaluation feedback form constructed in line with the evaluation questions:

1. Focus and quality of the RC's research

- Description of
  - the RC's research focus.
  - the quality of the RC's research (incl. key research questions and results)
  - the scientific significance of the RC's research in the research field(s)
- Identification of the ways to strengthen the focus and improve the quality of the RC's research

The additional material: TUHAT compilation of the RC's publications, analysis of the RC's publications data (provided by University of Leiden and the Helsinki University Library)
A written feedback from the aspects of: scientific quality, scientific significance, societal impact, innovativeness

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

2. Practises and quality of doctoral training

- Organising of the doctoral training in the RC. Description of the RC's principles for:
  - recruitment and selection of doctoral candidates
  - supervision of doctoral candidates
  - collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes
  - good practises and quality assurance in doctoral training
  - assuring of good career perspectives for the doctoral candidates/fresh doctorates
- Identification of the RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.

The additional material: TUHAT compilation of the RC's other scientific activities/supervision of doctoral dissertations
A written feedback from the aspects of: processes and good practices related to leadership and management

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

3. The societal impact of research and doctoral training

- Description on how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).
- Identification of the ways to strengthen the societal impact of the RC's research and doctoral training.

The additional material: TUHAT compilation of the RC's other scientific activities.
A written feedback from the aspects of: societal impact, national and international collaboration, innovativeness

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

4. International and national (incl. intersectoral) research collaboration and researcher mobility

- Description of
  - the RC's research collaborations and joint doctoral training activities
  - how the RC has promoted researcher mobility
- Identification of the RC's strengths and challenges related to research collaboration and researcher mobility, and the actions planned for their development.

A written feedback from the aspects of: scientific quality, national and international collaboration

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

5. Operational conditions

- Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).
- Identification of the RC's strengths and challenges related to operational conditions, and the actions planned for their development.

A written feedback from the aspects of: processes and good practices related to leadership and management

- Strengths
- Areas of development
- Other remarks
- Recommendations

6. Leadership and management in the researcher community

- Description of
  - the execution and processes of leadership in the RC
  - how the management-related responsibilities and roles are distributed in the RC
  - how the leadership- and management-related processes support
    - high quality research
    - collaboration between principal investigators and other researchers in the RC
    the RC's research focus
    - strengthening of the RC's know-how
- Identification of the RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes

7. External competitive funding of the RC

- The RCs were asked to provide information of such external competitive funding, where:
  - the funding decisions have been made during 1.1.2005-31.12.2010, and
  - the administrator of the funding is/has been the University of Helsinki
- On the e-form the RCs were asked to provide:

1) The relevant funding source(s) from a given list (Academy of Finland/Research Council, TEKES/The Finnish Funding Agency for Technology and Innovation , EU, ERC, foundations, other national funding organisations, other international funding organisations), and
2)The total sum of funding which the organisation in question had decided to allocate to the RCs members during 1.1.2005–31.12.2010.

Competitive funding reported in the text is also to be considered when evaluating this point.
A written feedback from the aspects of: scientific quality, scientific significance, societal impact, innovativeness, future significance

- Strengths
- Areas of development
- Other remarks
- Recommendations

8. The RC's strategic action plan for 2011–2013

- RC's description of their future perspectives in relation to research and doctoral training.

A written feedback from the aspects of: scientific quality, scientific significance, societal Impact, processes and good practices related to leadership and management, national and international collaboration, innovativeness, future significance

- Strengths
- Areas of development

- Other remarks
- Recommendations

9. Evaluation of the category of the RC in the context of entity of the evaluation material (1-8)

The RC's fitness to the chosen participation category
A written feedback evaluating the RC's fitness to the chosen participation category
- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

10. Short description of how the RC members contributed the compilation of the stage 2 material
Comments on the compilation of evaluation material

11. How the UH's focus areas are presented in the RC's research?
Comments if applicable

12. RC-specific main recommendations based on the previous questions 1–11

13. RC-specific conclusions

1.7 Evaluation criteria

The panellists were expected to give evaluative and analytical feedback to each evaluation question according to their aspects in order to describe and justify the quality of the submitted material. In addition, the evaluation feedback was asked to be pointed out the level of the performance according to the following classifications:

- outstanding (5)
- excellent (4)
- very good (3)
- good (2)
- sufficient (1)

Evaluation according to the criteria was to be made with thorough consideration of the entire evaluation material of the RC in question. Finally, in questions 1-4 and 9, the panellists were expected to classify their written feedback into one of the provided levels (the levels included respective descriptions, 'criteria'). Some panels used decimals in marks. The descriptive level was interpreted according to the integers and not rounding up the decimals by the editors.

Description of criteria levels

Question 1 – FOCUS AND QUALITY OF THE RC'S RESEARCH

Classification: Criteria (level of procedures and results)

Outstanding quality of procedures and results (5)

Outstandingly strong research, also from international perspective. Attracts great international interest with a wide impact, including publications in leading journals and/or monographs published by leading international publishing houses. The research has world leading qualities. The research focus, key research questions scientific significance, societal impact and innovativeness are of outstanding quality.

In cases where the research is of a national character and, in the judgement of the evaluators, should remain so, the concepts of "international attention" or "international impact" etc. in the grading criteria above may be replaced by "international comparability".

Operations and procedures are of outstanding quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are in alignment with the documentation. The ambition to develop the community together is of outstanding quality.

Excellent quality of procedures and results (4)

Research of excellent quality. Typically published with great impact, also internationally. Without doubt, the research has a leading position in its field in Finland.

Operations and procedures are of excellent quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of excellent quality.

Very good quality of procedures and results (3)

The research is of such very good quality that it attracts wide national and international attention.

Operations and procedures are of very good quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of very good quality.

Good quality of procedures and results (2)

Good research attracting mainly national attention but possessing international potential, extraordinarily high relevance may motivate good research.

Operations and procedures are of good quality, shared occasionally in the community. The improvement of research and other efforts are occasionally documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of good quality.

Sufficient quality of procedures and results (1)

In some cases the research is insufficient and reports do not gain wide circulation or do not have national or international attention. Research activities should be revised.

Operations and procedures are of sufficient quality, shared occasionally in the community. The improvement of research and other efforts are occasionally documented and operations and practices are to some extent in alignment with the documentation. The ambition to develop the community together is of sufficient quality.

Question 2 – DOCTORAL TRAINING
Question 3 – SOCIETAL IMPACT
Question 4 – COLLABORATION

Classification: Criteria (level of procedures and results)

Outstanding quality of procedures and results (5)

Procedures are of outstanding quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are in alignment with the documentation. The ambition to develop the community together is of outstanding quality. The procedures and results are regularly evaluated and the feedback has an effect on the planning.

Excellent quality of procedures and results (4)

Procedures are of excellent quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of excellent quality. The procedures and outcomes are evaluated and the feedback has an effect on the planning.

Very good quality of procedures and results (3)

Procedures are of very good quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and

management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of very good quality.

Good quality of procedures and results (2)

Procedures are of good quality, shared occasionally in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of good quality.

Sufficient quality of procedures and results (1)

Procedures are of sufficient quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are occasionally documented and operations and practices are to some extent in alignment with the documentation. The ambition to develop the community together is of sufficient quality.

Question 9 – CATEGORY

Participation category – fitness for the category chosen

The choice and justification for the chosen category below should be reflected in the RC's responses to the evaluation questions 1–8.

1. *The research of the participating community represents the international cutting edge in its field.*
2. *The research of the participating community is of high quality, but the community in its present composition has yet to achieve strong international recognition or a clear break-through.*
3. *The research of the participating community is distinct from mainstream research, and the special features of the research tradition in the field must be considered in the evaluation.* The research is of high quality and has great significance and impact in its field. However, the generally used research evaluation methods do not necessarily shed sufficient light on the merits of the research.
4. *The research of the participating community represents an innovative opening*. A new opening can be an innovative combination of research fields, or it can be proven to have a special social, national or international demand or other significance. Even if the researcher community in its present composition has yet to obtain proof of international success, its members can produce convincing evidence of the high level of their previous research.
5. *The research of the participating community has a highly significant societal impact.* The participating researcher community is able to justify the high social significance of its research. The research may relate to national legislation, media visibility or participation in social debate, or other activities promoting social development and human welfare. In addition to having societal impact, the research must be of a high standard.

An example of outstanding fitness for category choice (5) [5]

The RC's representation and argumentation for the chosen category were convincing. The RC recognized its real capacity and apparent outcomes in a wider context to the research communities. The specific character of the RC was well-recognized and well stated in the responses. The RC fitted optimally for the category.

- Outstanding (5)
- Excellent (4)
- Very good (3)
- Good (2)
- Sufficient (1)

The above-mentioned definition of outstanding was only an example in order to assist the panellists in the positioning of the classification. There was no exact definition for the category fitness.

[5] The panels discussed the category fitness and made the final conclusions of the interpretation of it.

1.8 Timetable of the evaluation

The main timetable of the evaluation:

1.	Registration	November 2010
2.	Submission of self-evaluation materials	January–February 2011
3.	External peer review	May–September 2011
4.	Published reports	March–April 2012
	- University level public report	
	- RC specific reports	

The entire evaluation was implemented during the university's strategy period 2010–2012. The preliminary results were available for the planning of the following strategy period in late autumn 2011. The evaluation reports will be published in March/April 2012. More detailed time schedule is published in the University report.

1.9 Evaluation feedback – consensus of the entire panel

The panellists evaluated all the RC-specific material before the meetings in Helsinki and mailed the draft reports to the evaluation office. The latest interim versions were on-line available to all the panellists on the Wiki-sites. In September 2011, in Helsinki the panels discussed the material, revised the first draft reports and decided the final numeric evaluation. After the meetings in Helsinki, the panels continued working and finalised the reports before the end of November 2011. The final RC-specific reports are the consensus of the entire panel.

The evaluation reports were written by the panels independently. During the editing process, the evaluation office requested some clarifications from the panels when necessary. The tone and style in the reports were not harmonized in the editing process. All the reports follow the original texts written by the panels as far as it was possible.

The original evaluation material of the RCs, provided for the panellists is attached at the end of the report. It is essential to notice that the exported lists of publications and other scientific activities depend how the data was stored in the TUHAT-RIS by the RCs.

2 Evaluation feedback

2.1 Focus and quality of the RC's research

- *Description of*
  - *the RC's research focus*
  - *the quality of the RC's research (incl. key research questions and results)*
  - *the scientific significance of the RC's research in the research field(s)*
- *Identification of the ways to strengthen the focus and improve the quality of the RC's research*

ASPECTS: Scientific quality, scientific significance, societal impact, innovativeness

Most of the studies during 2005-2010 performed by the teams in this RC relate to human molecular genetics and are based on collaboration between clinicians and strongly committed patients and families. The members of this RC have been very successful in using genomics tools to pinpoint gene mutations/alleles/loci for a set of diseases including lactase deficiencies, autism spectrum disorders and eye diseases. The group has been very productive in publishing peer-reviewed articles and produced during that period a low but appreciated number of PhD theses with quite limited resources as compared to other RCs with sometimes a ten-fold higher budget.

Three PhD theses have focused on gene identification of human lactase deficiencies, two PhD theses on genes involved in eye diseases and one PhD thesis on the genetics of autism. Impressively, over 100 papers have been produced mainly in the field of human genetics (which in general yields high impact factors) but the number of citations per publication (8.0) is not on the high end. Societal spin-offs of the research have been significant with the development of genetic test of lactase deficiency with over 10,000 tests being performed in Finland per year. In addition young scientists and/or doctors have been trained very well and have meanwhile returned to important scientific and/or societal (e.g. health care) positions.

The proposed new focus on an even more challenging task is to use current molecular and statistical genetics methods to elucidate the cross-talk between genes and environment in music perception and production. This is a highly complex task as music relates to many different biological aspects (perception, emotion, cognitivity, motoric skills). The major strength of this RC is that it proposes a unique and original plan to identify genetic variants affecting the ability to perceive music using the combination of family-based and quantitative-driven approaches with genomics. This approach is similar to previous studies with collection of families having specific phenotypes and aiming to identify common genetic denominators, and which has in their case proven successful for the diseases listed above. This exciting new "music-gene" work, which goes beyond disease and seems unlike other projects in recent times better supported, is done in collaboration with the Sibelius Academy and the University of Helsinki. The RC has the competence to carry out the work, certainly based on their publication record of the past, albeit in different fields. Coupling music and genetics is extremely interesting from many standpoints, including the evolutionary one, and it is quite obvious that any finding will have a high scientific impact and raise high interest with the lay public.

The concerns may of course be the number of families that are required to accurately identify the genes and alleles involved, although the RC has demonstrated that haplotypes (of AVPR1) are associated with music listening. Another question is the funding, which has been (too) low, but the added extra information tells that this music-genetics project is recently well funded. These studies tend to be quite expensive and even more pronounced when many families need to be analyzed for gene identification. For example, the international community has so far had only limited success in finding strong correlations between genotype and phenotype for multi-factorial diseases. Significant resources for carrying out the experiments as well as doing the analysis, and then validating the findings, are necessary. It is not fully clear whether this tantalizing RC has sufficient funding and families. There is a very high probability that the technical approach will need to be expanded. According to the study protocol GWAS and transcriptional profiling are indicated, yet NextGen sequencing will have to be used to identify low

frequent alleles. Another point is that we cannot fully understand how and which representative RNA samples can be retrieved from the families that are/will be part of the study. Another issue is whether such single project at this fairly preliminary stage (although interesting) fits to the category of RCs or should actually be part of another larger human genetics community project in order to have broader access to expertise and approach the proposed diverse lines of research.

Numeric evaluation: 4 (Excellent)

2.2 Practises and quality of doctoral training

- *Organising of the doctoral training in the RC. Description of the RC's principles for:*
  - *recruitment and selection of doctoral candidates*
  - *supervision of doctoral candidates*
  - *collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes*
  - *good practises and quality assurance in doctoral training*
  - *assuring of good career perspectives for the doctoral candidates/fresh doctorates*
- *Identification of the RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.*
- *Additional material: TUHAT compilation of the RC's other scientific activities/supervision of doctoral dissertations*

ASPECTS: Processes and good practices related to leadership and management

The RC, on the previous disease topics, has been reasonably productive in terms of PhD theses, and there are clear signs that the numbers in the context of these research teams may go up in the future. Six/seven PhDs were delivered during 2005-2010, and these PhDs have found new appropriate post-doc/permanent positions after dissertation. There are significantly more candidates than PhD positions, the RC has a keen eye on screening for important parameters (motivation, team spirit, …), and everything points to a strong recruitment position because of the attention that human molecular genetics currently has, in combination with the emerging reputation of the members of this RC.

PhD students are recruited by interviews and candidates are first employed for a test period of 3-6 months. As mentioned above, the ability of the selected young scientists to work together and be capable of the required networking between the different disciplines – including here art and science – are well monitored. The size of the RC is limited and supervision is done by everyday guidance and accompanied by an adequate course package.

Numeric evaluation: 4 (Excellent)

2.3 The societal impact of research and doctoral training

- *Description on how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).*
- *Identification of the ways to strengthen the societal impact of the RC's research and doctoral training.*
- *Additional material: TUHAT compilation of the RC's other scientific activities.*

ASPECTS: Societal impact, national and international collaboration, innovativeness

The RC has had extensive experience of societal interaction, which is what one may expect from a team in human genetics with a strong clinical component. The initial work on lactase deficiency has led to diagnostic kits that are used on a routine basis in clinical practice in Finland and elsewhere. This has improved diagnosis but also circumvented cumbersome gastroscopy. A cost-benefit analysis has also been done in collaboration with the Helsinki School of Economics and demonstrates the ambition of the RC taking a scientific finding into to use taking into the consideration the total cost for society.

The ongoing project to identify genetic variants linked to music talent and perception has wide implications for society and can possibly enable us better understand evolution of music and, by extension e.g. language, in human history. The study is a truly exciting example of the connection between art and science. Already the preliminary studies have given excellent press coverage as an indication of the interest to this topic.

Numeric evaluation: 5 (Outstanding)

2.4 International and national (incl. intersectoral) research collaboration and researcher mobility

- *Description of*
  - *the RC's research collaborations and joint doctoral training activities*
  - *how the RC has promoted researcher mobility*
- *Identification of the RC's strengths and challenges related to research collaboration and researcher mobility, and the actions planned for their development.*

ASPECTS: Scientific quality, national and international collaboration

The long list of national and international collaborators relates to previous and current research topics and demonstrates the opportunities for research mobility. However it is hard to estimate the extent of these interactions by the brief description. Even if the RC is limited in size it would be interesting to learn more about the planning and incitement of doctoral training abroad – is the purpose to bring expertise back into the group or is it more to export obtained knowledge? Are there any international collaborators on the proposed music topic and is there any need for complementary expertise, including genomics/genetics that could be sought through such collaborations?

Numeric evaluation: 4 (Excellent)

2.5 Operational conditions

- *Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).*
- *Identification of the RC's strengths and challenges related to operational conditions, and the actions planned for their development.*

ASPECTS: Processes and good practices related to leadership and management

Finnish research in the field of modern human genetics has a long tradition and is known worldwide. The current RC builds on that tradition of collecting Finnish families, which are known to be genetically more homogeneous than other populations and are therefore more suitable to carry out disease gene identification.

The success has also been linked to the access to an infrastructure for genotyping, sequencing and bio-informatics. It would be interesting to learn more about how the current developments in genomics are impacting the planned research and whether the current infrastructure has sufficient of capacity and instruments for the planned work.

Competence in bioinformatics is frequently limiting today's research and the new project may need a new set of computational expertise as the topic "music apture and perception" is likely significantly more complex than the previous projects due to its multi-factorial character. Are there sufficient competences within a core function of the UH or this entirely covered by the present group?

It is also unclear if the RC is planning for an international collaboration for linking music traits to genetics; this may be needed to confirm findings from the Finnish material and/or for seeking additional competences.

2.6 Leadership and management in the researcher community

- *Description of*
  - *the execution and processes of leadership in the RC*
  - *how the management-related responsibilities and roles are distributed in the RC*
  - *how the leadership- and management-related processes support*
    - *high quality research*
    - *collaboration between principal investigators and other researchers in the RC*
    - *the RC's research focus*
    - *strengthening of the RC's know-how*
- *Identification of the RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes*

ASPECTS: Processes and good practices related to leadership and management

The main PI of the RC has a solid background in molecular genetics combined with experience of clinical genetics. She has participated in appropriate leadership and pedagogical education. Since the RC is limited in size, little can be said about the leadership and organization of the RC.

A management plan that includes doctoral training, exchange programs and leadership training will be much more appropriate if the project is taken to the next phase such as establishing an international gene-music consortium.

2.7 External competitive funding of the RC

• The RCs were asked to provide information of such external competitive funding, where:
  • the funding decisions have been made during 1.1.2005–31.12.2010, and
  • the administrator of the funding is/has been the University of Helsinki

• On the e-form the RCs were asked to provide:
  1) The relevant funding source(s) from a given list (Academy of Finland/Research Council, TEKES/The Finnish Funding Agency for Technology and Innovation, EU, ERC, foundations, other national funding organisations, other international funding organizations), and
  2) The total sum of funding which the organisation in question had decided to allocate to the RCs members during 1.1.2005–31.12.2010.

Competitive funding reported in the text is also to be considered when evaluating this point.
ASPECTS: Scientific quality, scientific significance, societal impact, innovativeness and future significance

The external funding during the period 2005-2010 was very low (250 k €) and makes us feel worried given the ambitious plans for the RC. However, the PI indicates that funds have been secured for 2011 and beyond. To what extent and level these will suffice for this type of project is unclear. It was also noted that in the stage 1 material an EC grant was mentioned (1.7 M €), which is not part of the stage 2 material.

On the other hand over 100 papers were published during this period and 6-7 PhDs have still been produced. Thus the university funding must have been significant to allow the research to continue with the impressive scientific output.

Taken together, given the uncertainty of the funding situation it may be suggested that strategic alliances are also established for securing funding and allowing the exciting work to be carried out. For example, through other University of Helsinki groups, EC funding or BGI (China) could perhaps be more than happy to assist in the sequencing of samples if the scientific findings could lead to high impact paper(s) which is likely in this case.

2.8 The RC's strategic action plan for 2011–2013

• RC's description of their future perspectives in relation to research and doctoral training.
ASPECTS: Scientific quality, scientific significance, societal Impact, processes and good practices related to leadership and management, national and international collaboration, innovativeness, future significance

The strength of the action plan is that is describes a novel interdisciplinary approach using molecular genetics to approach the tantalizing topic of music production and perception. The literature in this field is very limited and the main contributions so far have been by the PI of this RC. Any finding would be highly interesting from many perspectives, both in science and society.

The action plan describes the combination of a doctoral training program that includes the Sibelius Academy, Medical Genetics and Statistical Genetics at the University of Helsinki. This is truly an interesting and novel constellation. However the approach used for going forward is not described in detail and there are many issues to address in order to be successful. These include additional funding, international collaborators, new recruitments or collaborators to solve the new issues relating to multiple genes, the technical approach given the new tools in genomics/sequencing, family sizes to identify low frequent alleles, recruitment of the families still, validation cohort etc.

2.9 Evaluation of the category of the RC in the context of entity of the evaluation material (1-8)

The RC's fitness to the chosen participation category.
Category 4. The research of the participating community represents an innovative opening.

The small RC is participating under category 4 (The research of the participating community represents an innovative opening) and this seems appropriate.

Numeric evaluation: 4 (Excellent)

2.10 Short description of how the RC members contributed the compilation of the stage 2 material

The main PIs contributed to most of the assembly of stage 2 material.

2.11 How the UH's focus areas are presented in the RC's research

Not selected focus area.
The RC is in line with the University of Helsinki's focus areas.

2.12 RC-specific main recommendations

The main recommendations are indicated in previous sections. The RC represents innovative research but need to address the following items.

- Funding – what is needed (extra) in terms of personnel and technical analysis?
- Establish (international) collaborations!
- Identify new recruitments or collaborators to address the more complex project.
- Decide on technical approach – outsource/collaborate.
- Recruitment of additional families – what is the required size?
- Plan for validation cohorts.

2.13 RC-specific conclusions

During the evaluation period 2005-2010, the main PI has been very successful, productive and efficient in terms of high publication record and high number of PhDs. The PI has also impressively demonstrated the ability of carrying out translational research by taking genetic findings into the routine clinical practice. In a drastic change of plan, the future research directions of the established RC now aim to address the genetics of musical perception and skill. This is a highly interesting project involving art, statistics and genetics, and is of a significantly more complex nature and hence challenge than the previous successful disease-orientated projects. The panel, confronted with successful reports from the past in disease-orientated projects followed by the drastic switch towards a challenging topic in this RC, doubts as to whether this is at present the best strategy for building an RC. In addition, the panel felt that this very ambitious project would better be carried out in a larger researcher community that has experience with the genetics of multi-factorial based phenotypes/diseases.

3 Appendices

A. Original evaluation material
    a. Registration material – Stage 1
    b. Answers to evaluation questions – Stage 2
    c. List of publications
    d. List of other scientific activities
B. Bibliometric analyses
    a. Analysis provided by CWTS/University of Leiden
    b. Analysis provided by Helsinki University Library (66 RCs)

International evaluation of research and doctoral training
at the University of Helsinki 2005-2010

RC-SPECIFIC MATERIAL FOR THE PEER REVIEW

NAME OF THE RESEARCHER COMMUNITY:
Gene-culture evolution in music (MUSGEN)

LEADER OF THE RESEARCHER COMMUNITY:
Professor Irma Järvelä, Department of Medical Genetics, Haartman-institute

RC-SPECIFIC MATERIAL FOR THE PEER REVIEW:

- Material submitted by the RC at stages 1 and 2 of the evaluation
  - STAGE 1 material: RC's registration form (incl. list of RC participants in an excel table)
  - STAGE 2 material: RC's answers to evaluation questions
- TUHAT compilations of the RC members' publications 1.1.2005-31.12.2010
- TUHAT compilations of the RC members' other scientific activities 1.1.2005-31.12.2010
- Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010 (analysis carried out by CWTS, Leiden University)

  NB! Since Web of Science(WoS)-based bibliometrics does not provide representative results for most RCs representing humanities, social sciences and computer sciences, the publications of these RCs will be analyzed by the UH Library (results available by the end of June, 2011)

INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI

RC-SPECIFIC STAGE 1 MATERIAL (registration form)

1 RESPONSIBLE PERSON

Name: Järvelä, Irma

E-mail:

Phone: 050 544 7030

Affiliation: University of Helsinki

Street address: Haartmaninkatu 8

2 DESCRIPTION OF THE PARTICIPATING RESEARCHER COMMUNITY (RC)

Name of the participating RC (max. 30 characters): Gene-culture evolution in music

Acronym for the participating RC (max. 10 characters): MUSGEN

Description of the operational basis in 2005-2010 (eg. research collaboration, joint doctoral training activities) on which the RC was formed (MAX. 2200 characters with spaces): The development of methods of quantitative genetics has facilitated the identification of genes affecting human complex traits. To understand human evolution during the last 100 000 years, it is of upmost importance to elucidate the role of cultural inheritance in biological evolution. What is the interplay between genetic mutation and social interaction leading to learning of new elements of culture? The music faculty is a multisensory trait that composes of different components like music perception and performance that may have different evolutionary histories and at molecular level are likely to be caused by different predisposing genetic variants. Based on our current understanding music integrates a wide variety of domains, cognitive, emotional, perceptual and motor but molecules mediating these effects remain so far uncharacterized. The proposed research protocol is highly interdisciplinary and represents an innovative opening where the modern methods of molecular and statistical genetics and bioinformatics and music education are combined to elucidate the cross-talk between genes and environment in music perception and production. To accomplish this, the expertise of the two universities is combined in doctoral training order to understand the biological underpinnings of music in human behavior. Joint doctoral training is carried out by experts in music education from Sibelius Academy that engages in research on performance and creative art and in molecular genetics from the Department of Medical genetics and in statistical genetics at the Department of Biology and Environmental Sciences, University of Helsinki. The project represents pioneer work at international and national level.

3 SCIENTIFIC FIELDS OF THE RC

Main scientific field of the RC's research: biological, agricultural and veterinary sciences

RC's scientific subfield 1: Genetics and Heredity

RC's scientific subfield 2: Evolutionary Biology

RC's scientific subfield 3: Mathematical and Computational Biology

RC-SPECIFIC STAGE 1 MATERIAL (registration form)

RC's scientific subfield 4: Music

Other, if not in the list: music education, neuropsychology

4 RC'S PARTICIPATION CATEGORY

Participation category: 4. Research of the participating community represents an innovative opening

Justification for the selected participation category (MAX. 2200 characters with spaces): We aim to identify genetic variants affecting the ability to perceive music using family based approach and genome wide scan and analyze the conservation of the identified variants in other species. This would give fundamental information about evolution of vocal learning and brain development of humans and other species. The Genome database provides us with over 600 species with full genome sequence (http://www.ncbi.nlm.nih.gov/Genomes/) that form an excellent resource for comparative genome studies. The proposed research protocol is highly interdisciplinary and represents an innovative opening both internationally and nationally. An understanding of the evolutionary history of music attempted in this study may reveal essential factors affecting human brain function and civilization. The advent of genome-wide sequence and polymorphisms available has offered tools to perform large-scale studies on human genetic variation and identify regions of positive selection. These data will be utilized in this study to identify regions of selection associated with musical aptitude and environmental factors contributing to musical activities i.e. music culture. A corresponding interdisciplinary effort has not been taken before. Furthermore, the effect of genes and environmental factors on musical aptitude and on traits related to practising music has not been analysed before. The study could lead to scientific breakthroughs in a diversity of research fields including molecular genetics and evolution sound and music, neurobiology of human brain and music therapy. It is expected that novel genetic variants contributing to the function of human brain, evolution of music and language in human history will be obtained. The study promotes innovation and cooperation between art and science and has wide societal impact.

5 DESCRIPTION OF THE RC'S RESEARCH AND DOCTORAL TRAINING

Public description of the RC's research and doctoral training (MAX. 2200 characters with spaces): The PI has supervised six PhD theses and six master's theses and reviewed two doctoral theses during 2005-2010. PI was responsible for teaching clinical genetics in medical school curriculum in 2010. Three PhD-theses focused on gene identification of human lactase deficiencies. The molecular background of human lactase deficiencies was dissected; DNA-variant associated with adult-type hypolactasia was located in a distal enhancer region 14kb upstream of the lactase gene whereas congenital lactase deficiency was caused by mutations in the coding region of the lactase gene itself. One PhD-thesis focused on molecular genetic studies on autism spectrum disorders (ASD). In this work the first genetic loci for Asperger syndrome were found. A total of 16 articles in collaboration with other autism research groups have been published based on the families collected from Finland. In eye diseases, two PhD-theses were finalized; they comprised the first molecular genetic studies on glaucoma, exfoliation syndrome and age-related macular (AMD) degeneration in Finland. The interaction between major risk genes in AMD modern methods of quantitative genetics and bioinformatics was demonstrated in 2008. In total 10 articles have been published in eye diseases. Related to children's brain development we have published five articles about novel disease and several novel mutations in X-chromosomal mental retardation (XLMR).

The first genome wide scan on musical aptitude in 15 Finnish families was published in 2008 showing that there is a genetic contribution to musical aptitude that is likely to be regulated by several predisposing genes or variants. As a sequel, the first candidate gene study in 2009 with the arginine vasopressin receptor 1A gene (AVPR1A) demonstrated an association with musical aptitude. The results suggest that musical aptitude is related to the neurobiological pathways affecting social affiliation and communication.

Significance of the RC's research and doctoral training for the University of Helsinki (MAX. 2200 characters with spaces): Identification of the SNP associated with LP/LNP was a major breakthrough in molecular genetics both internationally and nationally and obtained wide visibility in media. Later on, the group identified the founder mutation of LP/LNP in Arab Peninsula. The study had a profound effect on laboratory diagnosis of LP/LNP; genetic testing replaced traditional lactose tolerance test and improved the diagnosis of children's abdominal symptoms. Several lectures have been hold abroad and organized to educate Finnish physicians to perform genetic tests in primary health care. The study has had a significant impact on studies of human gene-culture evolution that is also the focus of the current proposal. The finding has facilitated cost-effective genetic testing of LP/LNP worldwide. Over 10 000 tests are being done every year in Finland. Based on the scientific findings the research group has introduced several other genetic tests into clinical practice. Noteworthy, during 2005-2010 the group has collected representative study materials for the aforementioned projects that are the prerequisite for future studies. The preliminary molecular genetic studies on musical aptitude that combined for the first time the fields of art and science were widely reviewed in international and national media. The PhDs of the group have been successful in their career; PhD Nabil Enattah from Libya after finalizing his PhD and working a total of eight years in Finland could transfer his knowledge he learnt to his home country. Mikko Kuokkanen has worked as post doc at the Broad Institute, Boston, USA, Tero Ylisaukko-oja works as a scientific adviser in a leading international pharmaceutical company, Susanna Lemmelä in a group financed by the Academy of Finland. Sanna Seitsonen MD PhD is training to become an ophthalmologist. During 2005-2010 the PI has been granted a total of 1.7 M € external money including an EU-grant for molecular genetics of ASD.

Keywords: gene, culture, SNP, association, evolution, musical aptitude, environment, education, interaction, expression

6 QUALITY OF RC'S RESEARCH AND DOCTORAL TRAINING

Justified estimate of the quality of the RC's research and doctoral training at national and international level during 2005-2010 (MAX. 2200 characters with spaces): Most of the studies performed by the group during 2005-2010 and earlier represent pioneer studies in human molecular genetics. The studies are based on close and long-standing collaboration between clinicians, patients and families who are willing to participate in the large projects. The PI and her group have proven the ability to productively establish new interdisciplinary approaches exemplified by the projects where genetic variant of lactase persistence/non-persistence was identified and the preliminary studies on genetic background of musical aptitude. The studies aim to open new avenues to study selection of human traits and genes and to understand human gene-culture evolution facilitated by the recent development of genomic and statistical approaches. The ability to perceive and produce music is a complex function of the human brain whose underlying biological basis remaining to be elucidated. The effort represents a novel interdisciplinary approach where molecules underlying artistic cognitive and their interaction with evolution of music culture will be elucidated. We aim

to answer the questions: What kind of effects the genome has on cultural evolution in music? Can music culture drive genetic evolution? Due to interdisciplinary character of the project the design of the studies and articles published from the project will be performed in close collaboration with experts in musical aptitude and music education in the group. Strong music culture and systematic education in music in Finland facilitates the study. The Finns have proven to be interested in the interdisciplinary project; more than 60 families with 700 members have given their DNA-samples to the study to date and the amount is increasing in the near future. The research group at the moment is small but will be expanded in the near future thanks to the funding obtained from the Academy of Finland for 2011-2014 and Biocentrum Helsinki for 2011-2013.

Comments on how the RC's scientific productivity and doctoral training should be evaluated (MAX. 2200 characters with spaces): During 2005-2010, 59 original articles and three reviews in international and six in domestic series have been produced by the group. Of them seven are published in high level journals of molecular genetics with IF around 10. Eight manuscripts are in process. A total of six PhD-theses and six masters's theses were finalized during 2005-2010. All these works were the first in their field in Finland for which extensive family and patient materials were collected. Genome wide scans of Asperger syndrome and exfoliation syndrome were pioneer works at the international level.

The group identified a novel syndrome of X-linked mental retardation and found novel mutations in

X-linked genes in Finnish families. The Hirsch index of the PI is currently 28. The data obtained will have wide applications in studies of the development of human brain, the neurobiological background of emotions, neurological and neuropsychiatric diseases and in the long run in the ability to develop new tools for therapy. The work will open new avenues for basic cell and neurobiological studies to study the function of the identified genes. The obtained results will be published in high level international scientific journals. The group main aim is to publish novel data and avoids not to replicate others findings in their work. As music culture and music industry have strong influence on society the group continues to distribute their findings to wide audience in the future. Interestingly, the preliminary results have obtained wide international recognition: http://www.sciencedaily.com/releases/2009/05/090526093925.htm

http://www.med.helsinki.fi/uutiset/20090525_UkkolaL.htm;
http://www.nature.com/embor/journal/v10/n12/pdf/embor2009241.pdf

www.discoverymagazine.com. TV-programs on biology underlying musical aptitude has been produced (in 2008, 2010). We have given numerous interviews in Finnish broadcasting company and in in international and domestic newspapers and magazines. In addition to scientific articles we will continue to write articles to educate teachers and students.

LIST OF RC MEMBERS

NAME OF THE RESEARCHER COMMUNITY:			MUSGEN		
RC-LEADER			I. Järvelä		
CATEGORY			4		
	Last name	First name	PI-status (TUHAT, 29.11.2010)	Title of research and teaching personnel	Affiliation
1	Järvelä	Irma	x	professor	Department of Medical Genetics
2	Raijas	Pirre		senior researcher	Sibelius Academy
3	Karma	Kai		professor	Sibelius Academy
4	Ukkola-Vuoti	Liisa		doctoral candidate	Department of Medical Genetics
5	Kantojärvi	Katri		doctoral candidate	Department of Medical Genetics
6	Ylisaukko-oja	Tero			Department of Medical Genetics
7	Seitsonen	Sanna			Department of Medical Genetics
8	Rasinperä	Heli			Department of Medical Genetics
9	Lemmelä	Susanna			Department of Medical Genetics
10	Kolho	Kaija-Leena	x	senior researcher	Hospital for Children and Adolescents, Helsinki University Central Hospital and University of Helsinki

INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI

RC-SPECIFIC STAGE 2 MATERIAL

BACKGROUND INFORMATION

Name of the RC's responsible person: Järvelä, Irma

E-mail of the RC's responsible person:

Name and acronym of the participating RC: Biological background of musical traits, MUSGEN

The RC's research represents the following key focus area of UH: -- Select --

Comments for selecting/not selecting the key focus area:

1 FOCUS AND QUALITY OF RC'S RESEARCH (MAX. 8800 CHARACTERS WITH SPACES)

- **Description of the RC's research focus, the quality of the RC's research (incl. key research questions and results) and the scientific significance of the RC's research for the research field(s).**

  The focus of the RC is identification of genetic factors and evolution affecting human diseases and traits. 1) We have dissected molecular background of human lactase deficiencies by showing that the common lactase persistence non-persistence trait (LP/LNP) is associated with single nucleotide polymorphisms (SNPs) in a distal enhancer region regulating the lactase gene (LCT) whereas the severe congenital type of lactase deficiency (CLD) is caused by mutations in the coding region of LCT. Identification of the single nucleotide polymorphism (SNP), cytosine (C) to thymidine (T) -13910 and the enhancer region of LCT associated with human LP/LNP lead to the breakthrough in molecular genetics. We demonstrated that C/T-13910 shows complete association with lactase activity in the intestinal wall and that the T-allele at -13910 locus regulates LCT at transcriptional level. The finding has established genetic testing in clinical practice worldwide and facilitated the identification of additional variants nearby in various ethnic groups. In total, we have found eight rare SNPs in the enhancer region in different ethnic groups: Fulani, Urban Saudi, Xhosa, mixed African and North-East Russian. Comparison of the haplotypes in the LCT region has revealed that the C/T-13910 variant of LP has occurred at least twice in human history. Association of lactase persistence on type 1 and 2 diabetes, ovarian cancer and prostate cancer has been excluded whereas the risk between LNP and colon cancer requires further studies. - Identification of the molecules affecting the normal down-regulation lactase activity during childhood is ongoing using genome-wide microarray expression and methylation studies. -In CLD, the group designed a genetic test to clinical diagnostics. Soon after, the RC identified CLD-mutations in Italian and Turkish families.

  2) Autism spectrum and mental retardation. About 2-3% of the population is affected by mental retardation. In autism, up to 80% of the patients become mentally retarded. During 2005-2010 the RC has identified causative mutations in four different genes (PHF8, PAK3, duplication of HUWE1-HADHA-genes and PORCN) in the Finnish families with X-linked mental retardation. Recently, using exome sequencing of X-chromosome four other genes in four families are under study, one of them a novel gene. Collection of new families with several affected members with mental retardation of unknown cause is underway. Autism spectrum disorders (ASD) are severe neuropsychiatric disorders characterized by stereotypic behavior, difficulties in social communication and language acquisition. Increasing prevalence of autism spectrum disorders (infantile autism and Asperger syndrome) with 1:150-1:200 has been found recently, the reason being unknown. Autism and ASD exhibit high heritability, although specific susceptibility genes still remain largely elusive. We have used genome-wide association (GWA) studies, candidate gene analyses and analysis of novel copy number variants (CNVs) to elucidate molecular background in ASD. GWA studies in AGRE and the Finnish sample set were combined in international collaboration that highlighted the importance of further analyses on 3p24-26 locus involving comprehensive molecular genetic analyses of oxytocin receptor gene (OXTR), a

functional candidate gene for autism. In the heterogeneity-based genome search meta-analysis (HEGESMA) of nine genome scans on autism or ASD region 7q22-q32 reached genome-wide significance both in weighted and un-weighted analyses. The RC participated in Specific Targeted project by the European Union where association analysis showed significant associations in MKL2 with ASD and between SND1 and autism in the Finnish and Northern Dutch populations, respectively. In addition, evidence for association was found in RELN and GRIK2. Autism often shows obsessive repetitive symptoms that are characteristic to obsessive-compulsive disorder (OCD). Aberrant glutamate function has been suggested to be a risk for both ASDs and OCD. A question about common genetic background of ASD and OCD was raised by autism genome project consortium (AGP) that found an association with rs1340513 in the JMJD2C gene at 9p24. The RC could replicate this finding in the Finnish sample set in the immediate vicinity of a SNP found to be associated with OCD between SLC1A1 and JMJD2C. Analysis of copy number variants (CNVs) in only 21 Finnish families resulted in the identification of a CNV in >20% of cases using Nimblegen 2.1M array. This is the highest number of CNVs found to date in autism.

3) Ophtalmological diseases. The RC has identified predisposing genes for most common ophthalmological diseases i.e. age-related macular degeneration (AMD) in the Finnish sample set and found evidence for gene-gene interaction between two major AMD associated loci CFH and LOC387715 using three methods, logistic regression, a synergy index and the mutual information (MI) index. The RC has performed the first genome wide scan (GWS) in exfoliation syndrome (ES) and found the first mutation in juvenile-onset glaucoma in the myocilin gene in an extended Finnish family. The RC has found the first mutation in the properdin gene in a large Finnish family with properdin deficiency.

4) Musical aptitude. To understand human evolution during the last 100 000 years, it would be outmost importance to elucidate the role of cultural inheritance in biological evolution. Is musical aptitude running in families due to favourable environment or genetic predisposition? Answers to these questions would clarify the picture of human evolution.The RC has performed the first genome wide scan (GWS) in the world to identify molecules associated with musical aptitude using variance component based statistical methods in 2008. Several loci were identified in 15 Finnish multigenerational families, of them most significant locus was found on chromosome 4q22 (LOD 3.33) and suggestive evidence of linkage at 8q13-21 (LOD 2.29) with the combined music test scores. Interestingly, a positive LOD score was found at 18q, a region previously linked to dyslexia (DYX6) using combined music test scores. The GWS on musical aptitude showed that there is a genetic contribution to musical aptitude that is likely to be regulated by several predisposing genes or variants. As a sequel, the first candidate gene study with the arginine vasopressin receptor 1A gene (AVPR1A) demonstrated an association with musical aptitude. Noteworthy, high heritability for creative functions of music (composing, arranging and improvising) were identified. Furthermore, high music test scores are significantly associated with creative functions in music. The work presented here represents pioneer work at international and national level. The effect of genes and environmental factors on musical aptitude and on traits related to practising music has not been analysed before. The results suggest that musical aptitude is related to the neurobiological pathways affecting social affiliation and communication that have a protective role in human evolution. Further studies using larger materials and tighter SNP-density (700 000K scan) and genome-wide expression array and methylation scan on opposite phenotypes in music perception and practice will give unique information about gene-culture co-evolution in music in the near future.

- **Ways to strengthen the focus and improve the quality of the RC's research.**

  We aim to utilize the modern methods of quantitative genetics and bioinformatics to facilitate the identification of genes affecting human complex traits. We use exome sequencing and copy number variant analysis to dissect genetic background underlying mental retardation and ASD. We perform genome wide association and gene expression, microRNA and methylation analysis in unique family material of musical aptitude to identify genes and/or genetic variants. Genome sequence of different

species available in databases (http://www.ncbi.nlm.nih.gov/Genomes/) will be used to analyse the conservation of the identified genes in other species. This would give fundamental information about evolution of vocal learning and brain development of humans and other species. Our study on the interaction of genetic variants and the environment in the most fascinating and common trait among humans, music, is groundbreaking and unique both internationally and nationally.

2 PRACTISES AND QUALITY OF DOCTORAL TRAINING (MAX. 8800 CHARACTERS WITH SPACES)

- How is doctoral training organised in the RC? Description of the RC's principles for recruitment and selection of doctoral candidates, supervision of doctoral candidates, collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes, good practises and quality assurance in doctoral training, and assuring good career perspectives for the doctoral candidates/fresh doctorates.

Seven PhD theses and six master's theses were completed and reviewed two doctoral theses during 2005-2010 in the RC. PI is responsible for teaching medical and clinical genetics in medical school curriculum starting in spring 2010. - Three PhD-theses focused on gene identification of human lactase deficiencies where the molecular background of both human lactase deficiencies was dissected. One PhD thesis (supervised in collaboration with by K-L Kolho) focused on the applicability of the SNP test in primary health care. One PhD-thesis focused on molecular genetic studies on autism spectrum disorders (ASD). In this work the first genetic loci for Asperger syndrome were found. A total of 16 articles in collaboration with other autism research groups both internationally and nationally have been published based on the families collected from Finland. In eye diseases, two PhD-theses were finalized; they comprised the first molecular genetic studies on glaucoma, exfoliation syndrome and age-related macular (AMD) degeneration in Finland. The interaction between major risk genes in AMD modern methods of quantitative genetics and bioinformatics was demonstrated in 2008. In total 10 articles have been published in eye diseases. Related to children's brain development we have published a novel duplication and novel mutations in three known genes (PHF8, PAK3, PORCN) in X-chromosomal disorders.

The research field of the RC is very attracting. There are a lot of applicants from both Finland and abroad who contact the RC and express their interest to join the group. Thus, there are more students interested that the RC can employ. The principal investigator chooses the candidates by personal interviews. The emphasis is put on high motivation and enthusiasm towards the project, good social skills and willingness to work efficiently. Suitable candidates are employed for a test period of 3- 6 months that is a test period both for the candidate and the existing group working together. Supervision of doctoral candidates takes place by everyday guidance, informal meetings, and e-mails to ensure fluent progress of the work. PI teaches and helps to write the first scientific article. The aim is to direct the student gradually to independent writing. Here academic writing courses organized by the University of Helsinki are helpful and recommended to each student. Collaborations with the Department of Biological and Environmental Sciences in statistical genetics, Hospital of Children and Adolescents, University of Helsinki, Rinnekoti Rehabilitation Institute, Jyvaskyla Central Hospital in recruiting patient materials are long-standing. Close collaboration with Sibelius Academy has been established in the project where biological background of musical traits are being studied. The aim of this interdisciplinary project is to combine the modern methods of molecular genetics and music education to promote collaboration between science and art.

Doctoral students have participated annually in international and domestic meetings in their fields. The aim of the RC is to do internationally remarked top quality science and publish articles in respected well-known series. The doctoral candidates/fresh doctorates are encouraged to apply to distinguished laboratories abroad for post-doctoral training.

Doctorates from the RC community have obtained good positions; PhD Tero Ylisaukko-oja is scientific adviser in large international pharmaceutical company, PhD Mikko Kuokkanen moved to Broad Institute, Boston for post doc, PhD Susanna Lemmelä obtained a post-doc position in a group financed by the Academy of Finland, MD PhD Sanna Seitsonen is graduating as an ophthalmologist. MD PhD Nabil Enattah moved back to his home country Libya where he utilizes his knowledge by educating the students in molecular genetics. MD, PhD Sari Anthoni is a chief physician in a distinguished private medical clinic.

- RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.

  The strengths of the RC are the interdisciplinary nature of the research. Collaboration with both Helsinki School of Economics and Sibelius-Academy has been fruitful. The students are trained in multidisciplinary atmosphere that is inspiring and increases creativity. The challenge is that multidisciplinary approach is more demanding and requires good networking skills. However, this kind of crosstalk is innovative an promotes new ideas between science and art and will be continued by the RC in the future. Maybe self-evidently, the group combines the clinical medicine, biochemical data and molecular genetic data to educate the students.

3 SOCIETAL IMPACT OF RESEARCH AND DOCTORAL TRAINING (MAX. 4400 CHARACTERS WITH SPACES)

- Description of how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).

  Identification of the variants of human lactase deficiencies has had a profound effect on clinical practice when facilitating genetic testing of LP/LNP. Genetic testing of LP/LNP and CLD have replaced inaccurate and tedious lactose tolerance test (LTT) in general practice and specialist clinics worldwide. Similarly, genetic test of CLD has replaced invasive and expensive gastroscopy also in case of CLD in newborns. In collaboration with the Helsinki School of Economics we have shown that genetic testing is cost-effective when saving time of the patient and laboratory staff.

  The research protocol is highly interdisciplinary and represents an innovative opening both internationally and nationally. An understanding of the evolutionary history of music attempted in this study may reveal essential factors affecting human brain function and civilization. The advent of genome-wide sequence and polymorphisms available has offered tools to perform large-scale studies on human genetic variation and identify regions of positive selection. These data will be utilized in this study to identify regions of selection associated with musical aptitude and environmental factors contributing to musical activities i.e. music culture. A corresponding interdisciplinary effort has not been taken before. Furthermore, the effect of genes and environmental factors on musical aptitude and on traits related to practising music has not been analysed before. The study can lead to scientific breakthroughs in a diversity of research fields including molecular genetics and evolution sound and music, neurobiology of human brain and music therapy. It is expected that novel genetic variants contributing to the function of human brain, evolution of music and language in human history will be obtained. The study promotes innovation and cooperation between art and science and has wide societal impact.

  Identification of genetic variants and mutations in autism spectrum disorders and mental retardation

  will produce new data about human brain function, helps in diagnostics and genetic counselling: One of the major goals in disease studies are the development of new genetic test into clinical practice and to apply bioinformatics tools to study gene-gene and gene-environment (culture) interaction.

Molecular genetic studies on musical traits have attracted international media.

Public citations of our work:

http://www.sciencedaily.com/releases/2009/05/090526093925.htm

http://discovermagazine.com/2009/jan/052/?searchterm=genes%20musical%20abilities

http://www.med.helsinki.fi/uutiset/20090525_UkkolaL.htm

http://www.nature.com/embor/journal/v10/n12/pdf/embor2009241.pdf

http://tv1.yle.fi/ PRISMA TV-program Biology underlying musical aptitude in 2008 and 2010

Several interviews in Finnish broadcasting company, articles in domestic newspapers and magazines.

- Ways to strengthen the societal impact of the RC's research and doctoral training.

  The aim of the RC is to increase the visibility of the research by writing popular articles in newspapers, by participating in TV- programs and broadcasting. The emphasis is to distribute the knowledge about the interaction of genes and culture in human diseases and traits to wide audience.

4 INTERNATIONAL AND NATIONAL (INCL. INTERSECTORAL) RESEARCH COLLABORATION AND RESEARCHER MOBILITY (MAX. 4400 CHARACTERS WITH SPACES)

- Description of the RC's research collaborations and joint doctoral training activities and how the RC has promoted researcher mobility.

  National and international collaborators and their impact to the project

  Professor Guy Froyen, University of Leuven, Belgium, Identification of genes underlying X-linked mental retardation

  FP6 AUTISM MOLGEN Strategic Targeted Research Project #512128 co-ordinated by Professor Anthony Bailey, University of Oxford. European autism materials were combined to identify predisposing genes for autism and found a new gene association using the Finnish sample set.

  Professor Kai Karma, Sibélius Academy, Helsinki, Finland, collecting and testing families

  Doctor of Music Pirre Raijas, Sibélius Academy, Helsinki, Finland, collecting and testing families

  Dr Harald H Göring, Department of Genetics, Southwest Foundation for Biomedical Research, San Antonio, TX, USA, variance-component based linkage analyses, advice and help in checking the results

  Associate professor Päivi Onkamo, University of Helsinki, help and advice in statistical genetic analyses

  Professor Brian Meyers, King Faisal Hospital, Riadh, Saudi-Arabia: the founder DNA-variant of lactose intolerance identified in Urban Saudi population

  Professor M Iqbal Parker, lactase persistence variants in African populations, University of

  Cape Town, South Africa

  Professor Ilkka Immonen, Department of Ophthalmology, University of Helsinki, molecular genetics of age related macular degeneration (AMD)

  MD Eva Forsman, Folkhälsan Research Institute, Helsinki, molecular genetics of glaucoma and exfoliation syndrome

  Professor Henrik Forsius, Folkhälsan Research Institute, Helsinki, molecular genetics of glaucoma and exfoliation syndrome

Dr. Lakshmi Muthuswamy, Ontario Cancer Research Institute, Toronto, autism copy number variants

Adjuct professor Vera Kalscheuer, Max Planck Institute, Berlin, exome sequencing

Adjuct professor Gajja Salomons, VU University Medical Center, Amsterdam, XLMR

MD PhD Aki Mustonen, Oulu University hospital, Finland, clinical diagnosis of mental retardation

MD Maarit Peippo, Family Welfare Federation, Helsinki, clinical diagnosis of mental retardation, Docent Mirja Somer, Family Welfare Federation, Helsinki, clinical diagnosis of mental retardation

MD Raija Vanhala, Lasten ja Nuorten sairaala, clinical diagnosis in autism spectrum disorders

LT Kristiina Avela, Family Welfare Federation, Helsinki, clinical diagnosis of mental retardation Professor Erkki Savilahti, clinical and biochemical analyses of lactase deficiencies

Adjunct Professor Kaija-Leena Kolho, clinical diagnosis of lactase deficiencies

Professor Teemu Malmi, Helsinki School of Economics, cost-effectiveness of genetic testing

The research mobility of the students:

PhD Mikko Kuokkanen carried out his post doc at the Broad Institute, Boston

PhD Tero Ylisaukko-oja works as a scientific adviser in a leading international pharmaceutical company

PhD Susanna Lemmelä works as a post doc financed by the Academy of Finland

MD Mari Korpela (nee Auranen) is graduated as a neurologist

MD Sanna Seitsonen graduates as an opththalmologist

MD, PhD Sari Anthoni, chief physician, private medical company

- RC's strengths and challenges related to research collaboration and researcher mobility, and the actions planned for their development.

The RC is characterized by wide interests in their research on molecular genetics. They have carried out extensive international scientific collaboration with numerous groups (collaborators mentioned above). Mobility of the students has been excellent. Post-doctoral position has been found easily for those willing to move abroad. It is still a challenge for post-doctoral students to find a post in Finland due to limited number of positions and limited amount of money for these positions in this country. This is the case for example with bioinformatics, a growing field working in close relationship with molecular genetics (wet lab).

5 OPERATIONAL CONDITIONS (MAX. 4400 CHARACTERS WITH SPACES)

- Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).

The RC works at the Department of Medical Genetics, Haartman Institute that has been the pioneer in molecular genetic studies of human diseases in Finland. It has established it's position as one of the leading institutes in the field and is one of the internationally most recognized and successful Department at the Faculty of Medicine and the University of Helsinki in scientific merits. Long traditions and established position offer a good working environment, located in new Biomedicum1 building at the Meilahti Campus. Most of the clinical collaborators work in closely located Helsinki University Hospital and Family Welfare Federation.

The Department of Medical Genetics has the equipments to perform the studies presented here. Biomedicum Helsinki provides an excellent infrastructure for the work laboratory and the biocomputing parts of the effort. Core facilities exist for STR and SNP genotyping, sequencing http://www.bioinfo.helsinki.fi/SeqLab/), and biocomputing http://www.bioinfo.helsinki.fi/).

The PI of the RC is responsible for teaching medical and dental students. In Medical genetics typically the gap between scientific findings and clinical diagnostics is narrow, the teaching can be relatively easily designed to cover topics. The other group members participate in teaching only rarely thus being able to concentrate on research.

- RC's strengths and challenges related to operational conditions, and the actions planned for their development.

The study materials in this study present genetically well-characterized homogeneous Finnish population that offer a unique possibility to identify genetic variants associated with diseases and traits compared to mixed populations. This is further strengthened by homogeneous Finnish culture due to geographical and cultural isolation. It can be hypothesized that rare variants with major effect are enriched in the isolated population. This can be the case with rare mutations in brain diseases. The setting further facilitates gene-gene and gene-culture interaction studies in multifactorial traits like musical aptitude. - So far the costs of new molecular genetic techniques including exome and whole genome sequencing are high and thus for the identification of genetic variants international collaboration has been carried out by the RC. In cases when laboratory services are bought competitive bidding has been and must be organized between top quality laboratories to save money.

6 LEADERSHIP AND MANAGEMENT IN THE RESEARCHER COMMUNITY (MAX. 4400 CHARACTERS WITH SPACES)

- Description of the execution and processes of leadership in the RC, how the management-related responsibilities and roles are distributed in the RC and how the leadership- and management-related processes support high quality research, collaboration between principal investigators and other researchers in the RC, the RC's research focus and strengthening of the RC's know-how.

The principal investigator has acquired specialist degrees both in general practice in 1986 and in clinical genetics in 1996 and has been appointed as adjunct professor in medical molecular genetics in 1996 at the University of Helsinki. She has over 10 years' experience in clinical diagnostics and over 20 years' experience in molecular genetic studies on human diseases and traits. She has performed her post-doctoral studies at the University College London Medical School in 1992-1994 where the work performed resulted in a total of eight high level original articles. The post-doc period culminated in the identification of the novel CLN3 gene underlying the most common progressive brain disorder in childhood published in Cell. In addition, PI has educated herself in leadership skills by participating in Leadership Education Program for Health Care (TeJOKO), at the Helsinki School of Economics in 2002-2003. To educate herself in teaching skills the PI has

has participated in Course in University Pedagogy for Medical Teachers in 2005-06, Course in University Pedagogy for Medical Teachers II in 2007-08 and Course in problem based learning (PBL) in 2008. The most important positions held by PI are acting as senior scientist, at the National Public Health Institute during 1995-2000 when she started her own group, clinical geneticist responsible for genetic testing and the implementation of new genetic tests into clinical practice during 2000-2005. In 2006 PI worked as a senior Scientist at the Academy of Finland. Since 2010 PI has been working as the acting professor, at the Department of Medical Genetics, University of Helsinki.

RC-SPECIFIC STAGE 2 MATERIAL

- RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes.

  To improve leadership and management skills the PI will participate in courses organized by the University of Helsinki to follow the strategic goals of the university. To develop the academic skills the RC will regularly participate in international meetings including those on human genetics, bioinformatics and statistical genetics.

7 EXTERNAL COMPETITIVE FUNDING OF THE RC

- Listing of the RCs external competitive funding, where:
  - the funding decisions have been made during 1.1.2005-31.12.2010, and
  - the administrator of the funding is/has been the University of Helsinki

- Academy of Finland (AF) - total amount of funding (in euros) AF has decided to allocate to the RC members during 1.1.2005-31.12.2010: 250000

- Finnish Funding Agency for Technology and Innovation (TEKES) - total amount of funding (in euros) TEKES has decided to allocate to the RC members during 1.1.2005-31.12.2010: 0

- European Union (EU) - total amount of funding (in euros) EU has decided to allocate to the RC members during 1.1.2005-31.12.2010: 0

- European Research Council (ERC) - total amount of funding (in euros) ERC has decided to allocate to the RC members during 1.1.2005-31.12.2010: 0

- International and national foundations – names of international and national foundations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
  - names of the foundations:
  - total amount of funding (in euros) from the above-mentioned foundations:

- Other international funding - names of other international funding organizations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
  - names of the funding organizations:
  - total amount of funding (in euros) from the above-mentioned funding organizations:

- Other national funding (incl. EVO funding and Ministry of Education and Culture funded doctoral programme positions) - names of other national funding organizations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
  - names of the funding organizations:
  - total amount of funding (in euros) from the above-mentioned funding organizations:

RC-SPECIFIC STAGE 2 MATERIAL

8 RC'S STRATEGIC ACTION PLAN FOR 2011–2013 (MAX. 4400 CHARACTERS WITH SPACES)

- Description of the RC's future perspectives in respect to research and doctoral training.

The development of methods of quantitative genetics and sequencing have facilitated the identification of genes affecting human diseases and traits. To understand human evolution during the last 100 000 years, it is of upmost importance to elucidate the role of cultural inheritance in biological evolution. The RC:s experience obtained in molecular genetics of human brain disorders and lactase deficiencies with interesting gene-culture co-evolution questions will be further utilized in the project to elucidate predisposing genes for musical aptitude. What is the interplay between genetic mutation and social interaction leading to learning of new elements of culture? The music faculty is a multisensory trait that composes of different components like music perception and performance that may have different evolutionary histories and at molecular level are likely to be caused by different predisposing genetic variants. Based on our current understanding music integrates a wide variety of domains, cognitive, emotional, perceptual and motor but molecules mediating these effects remain so far uncharacterized. The proposed research protocol is highly interdisciplinary and represents an innovative opening where the modern methods of molecular and statistical genetics and bioinformatics and music education are combined to elucidate the cross-talk between genes and environment in music perception and production. To accomplish this, the expertise of the two universities is combined in doctoral training order to understand the biological underpinnings of music in human behavior. Joint doctoral training is carried out by experts in music education from Sibelius Academy that engages in research on performance and creative art and in molecular genetics from the Department of Medical genetics and in statistical genetics at the Department of Biology and Environmental Sciences, University of Helsinki. The project represents pioneer work at international and national level. The study can lead to scientific breakthroughs in a diversity of research fields including molecular genetics, neurobiology and music education and therapy. It is expected that the study will give novel information about genes underlying human brain function, social communication and the evolution of music and language in human history. It is expected that novel genes and metabolic networks will be identified. The data obtained will have wide applications in studies of the development of language, the neurobiological background of emotions, neurological and neuropsychiatric diseases and in the long run to develop new tools for music therapy. The work will open new avenues for basic cell and neurobiological studies to study the function of the identified genes.

The effort represents a novel interdisciplinary approach where molecules underlying artistic cognitive and their interaction with evolution of music culture will be elucidated. It is a virgin territory to be explored.

The obtained results will be published in high level international scientific journals.

9 SHORT DESCRIPTION OF HOW THE RC MEMBERS HAVE CONTRIBUTED TO THE COMPILATION OF THE STAGE 2 MATERIALS (MAX. 1100 CHARACTERS WITH SPACES).

PI has collected the materials on stage 2 and written the major part of the text. Co-PI Kaija-Leena Kolho has written the text on her part. PhD-student Liisa Ukkola-Vuoti has read the text.

7 External competitive funding of the RC

External competitive funding obtained by the name of PI, the funding decisions made during 1.1.2005-31.12.2010 is shown below. The funding administered by the University of Helsinki is marked as bold.

Years	Funding agency	Sum €	Project
2005-2007	**The Academy of Finland**	**150000**	**Molecular genetics of autism spectrum disorders**
2006	**The Academy of Finland**	**103900**	**Molecular genetics of human lactase deficiencies**
2005-2007	The Sigrid Jusélius Foundation	128000	Molecular genetics of human lactase deficiencies
2005-2006	The Finnish Cultural Foundation	90000	Molecular genetics of musical aptitude
2005-2007	Helsinki University Hospital	180000	Molecular genetics of human lactase deficiencies
2007-2011	Helsinki University Hospital	150 000	Molecular genetics of autism spectrum disorders and mental retardation
2006-2008	European Union Specific Targeted project	278000	Using European and International populations to Identify Autism Susceptibility Loci
2006	The Paulo Foundation	8000	Genetics of musical aptitude
2008	The Nissi Foundation	10000	Genetics of glaucoma and exfoliation syndrome
2010	The Sigrid Jusélius Foundation	50000	Molecular genetics of autism and mental retardation
2005-2011	Finnish Medical Society	94800	Molecular genetics of autism spectrum disorders and glaucoma
2011-2014	**The Academy of Finland**	**948860**	**Biological basis of musical aptitude**
2011-2013	**Biocentrum Helsinki**	**144000**	**Biological basis of musical aptitude**

RC-member A Liisa Ukkola 35 000 € The Helsinki University Research Foundation, The Paulo Foundation
RC member B Katri Kantojärvi 6000 € The Rinnekoti Research Foundation
RC member C Mikko Kuokkanen 16 000 € The Finnish Cultural Foundation
RC member D Anne M Koivisto 2 400€ The Rinnekoti Research Foundation
RC member E Heli Rasinperä 17 000 € The Helsinki University Research Foundation, The Ida Montin Foundation
RC member F Susanna Lemmelä 4 000 € The Päivikki and Sakari Sohlberg Foundation
RC member G Sari Anthoni 5 000 € The Foundation for Allergy Research, Helsinki

Gene-culture co-evolution in music
RC: PI Professor Irma Järvelä
The Department of Medical Genetics, University of Helsinki Finland
http://www.helsinki.fi/haartman/lgo/tutkimus/music/english.htm

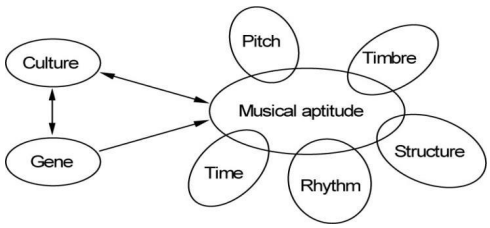

Figure1. The premise of the study is that genetic variants are associated with musical aptitude expressed as multiple traits. Musical aptitude has shaped the culture and cultural practices have affected the human genome and musical aptitude.

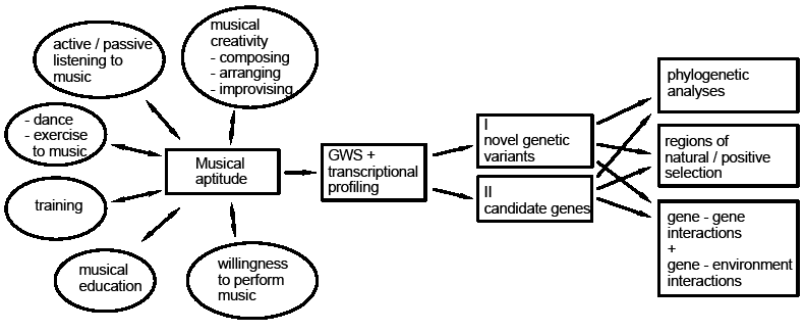

Figure 2**.** Schematic presentation of the **s**tudy protocol.

INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI

RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

MUSGEN/Järvelä

1 Analysis of publications

- Associated person is one of Irma Järvelä , Liisa Ukkola-Vuoti , Sanna Seitsonen , Heli Anna Emilia Rasinperä , Kaija-Leena Kolho ,

	Publication year						
Publication type	**2005**	**2006**	**2007**	**2008**	**2009**	**2010**	**Total Count 2005 - 2010**
A1 Refereed journal article	20	13	23	18	19	21	114
A2 Review in scientific journal			1		2		3
A4 Article in conference publication (refereed)						1	1
B1 Unrefereed journal article	5	2	6	4	3		20
D1 Article in professional journal						3	3
D2 Article in professional hand or guide book or in a professional data system, or text book material					2		2
E1 Popular article, newspaper article					1		1

2 Listing of publications

A1 Refereed journal article

2005

Bister, V, Kolho, K, Karikoski, R, Westerholm-Ormio, M, Savilahti, E, Saarialho-Kere, U **2005**, 'Metalloelastase (MMP-12) is upregulated in the gut of pediatric patients with potential celiac disease and in type 1 diabetes', **Scandinavian Journal of Gastroenterology**, vol 40, pp. 1413-1422.

Enattah, NS, Pekkarinen, T, Välimäki, M, Löyttyniemi, E, Järvelä, I, Enattah, NS **2005**, 'Genetically defined adult-type hypolactasia and self-reported lactose intolerance as risk factors of osteoporosis in Finnish postmenopausal women', **European Journal of Clinical Nutrition**, vol 59, no. 10, pp. 1105-1111.

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A4 Article in conference publication (refereed)

2010

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2005

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D1 Article in professional journal

2010

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2009

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2009

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INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI

RC-SPECIFIC TUHAT COMPILATIONS OF OTHER SCIENTIFIC ACTIVITIES 2005-2010

MUSGEN/Järvelä

1 Analysis of activities 2005-2010

- Associated person is one of Irma Järvelä , Liisa Ukkola-Vuoti , Sanna Seitsonen , Heli Anna Emilia Rasinperä , Kaija-Leena Kolho ,

Activity type	Count
Supervisor or co-supervisor of doctoral thesis	10
Participation in radio programme	5
Participation in TV programme	3

2 Listing of activities 2005-2010

Supervisor or co-supervisor of doctoral thesis

Irma Järvelä ,

Doctoral thesis supervision/Enattah, Irma Järvelä, 1997 → 2005, Finland

Doctoral thesis supervision /Ylisaukko-oja, Irma Järvelä, 2000 → 2005, Finland

Doctoral thesis supervision /Kuokkanen, Irma Järvelä, 2001 → 2006, Finland

Doctoral thesis supervision / Lemmelä, Irma Järvelä, 2002 → 2009, Finland

Doctoral thesis supervision /Rasinperä, Irma Järvelä, 2002 → 2006, Finland

Doctoral thesis supervision / Seitsonen, Irma Järvelä, 2004 → 2008, Finland

Doctoral thesis supervision /Kantojärvi, Irma Järvelä, 2007 → ..., Finland

Doctoral thesis supervision /Ukkola-Vuoti, Irma Järvelä, 2007 → ..., Finland

Doctoral thesis supervision, Irma Järvelä, 2010 → ..., Finland

Doctoral thesis supervision /Doagu, Irma Järvelä, 2010 → ..., Finland

Participation in radio programme

Irma Järvelä ,

Esiintyminen radio-ohjelmassa, Irma Järvelä, 19.05.2008, Finland

Esiintyminen radio-ohjelmassa, Irma Järvelä, 03.06.2009, Finland

Esiintyminen radio-ohjelmassa, Irma Järvelä, 03.03.2010, Finland

radiohaastattelu, Irma Järvelä, 16.10.2010

radiohaastattelu, Irma Järvelä, 17.10.2010

Participation in TV programme

Irma Järvelä ,

Esiintyminen TV-ohjelmassa, Irma Järvelä, 19.04.2008, Finland

Esiintyminen TV-ohjelmassa, Irma Järvelä, 03.02.2010, Finland

TV-haastettelu, Irma Järvelä, 17.10.2010

INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING
AT THE UNIVERSITY OF HELSINKI

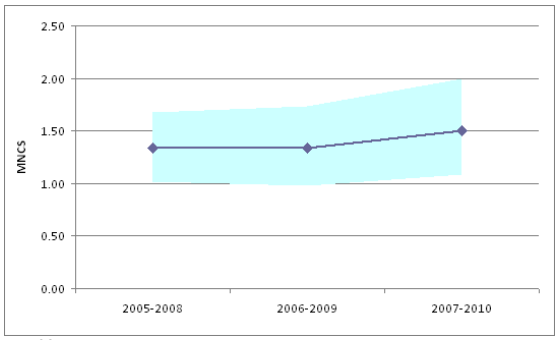
Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010
by CWTS, Leiden University, the Netherlands

Research Group: Järvelä I

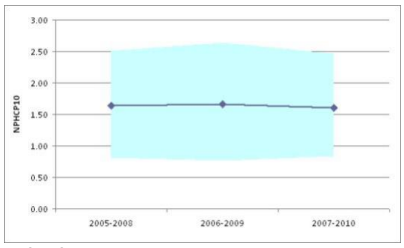
Basic statistics

Number of publications (P)	100
Number of citations (TCS)	780
Number of citations per publication (MCS)	8.04
Percentage of uncited publications	18%
Field-normalized number of citations per publication (MNCS)	1.36
Field-normalized average journal impact (MNJS)	1.16
Field-normalized proportion highly cited publications (top 10%)	1.48
Internal coverage	.90

Trend analyses



MNCS



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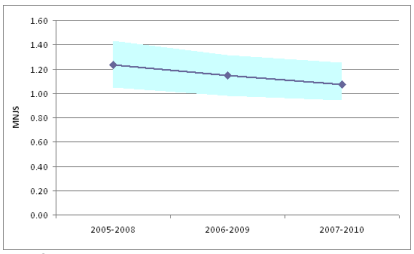
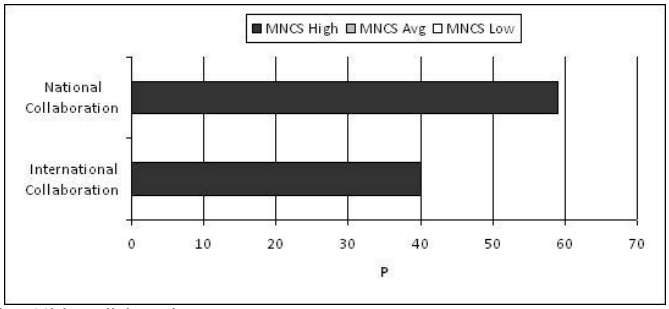


MNJS

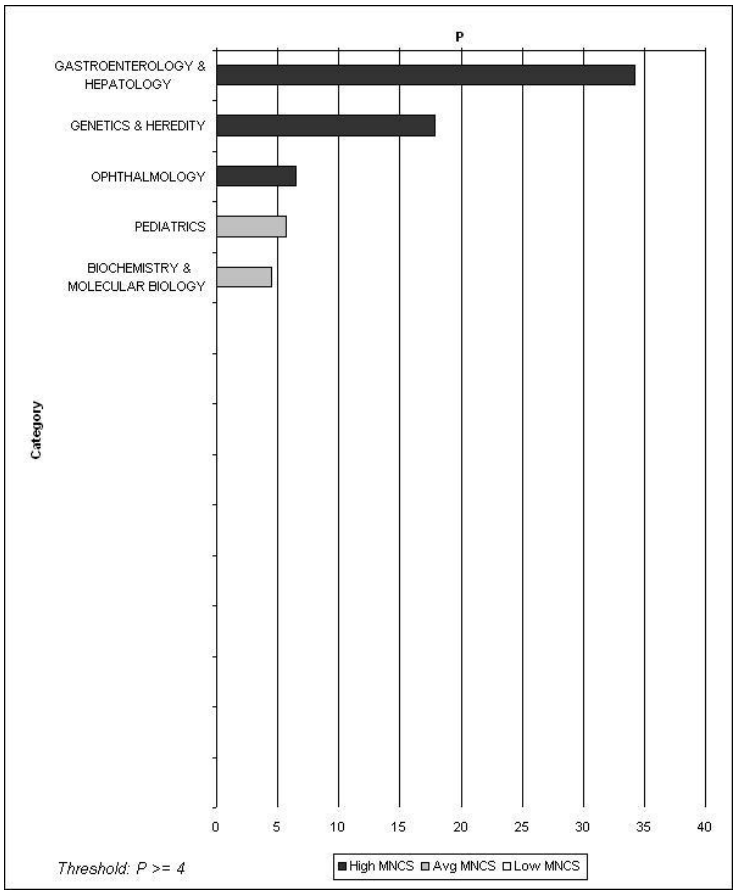
Collaboration



Performance (MNCS) by collaboration type

Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010
by CWTS, Leiden University, the Netherlands

Research profile



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